

CHAPTER 6

SMALL POPULATIONS AND GENETIC DRIFT

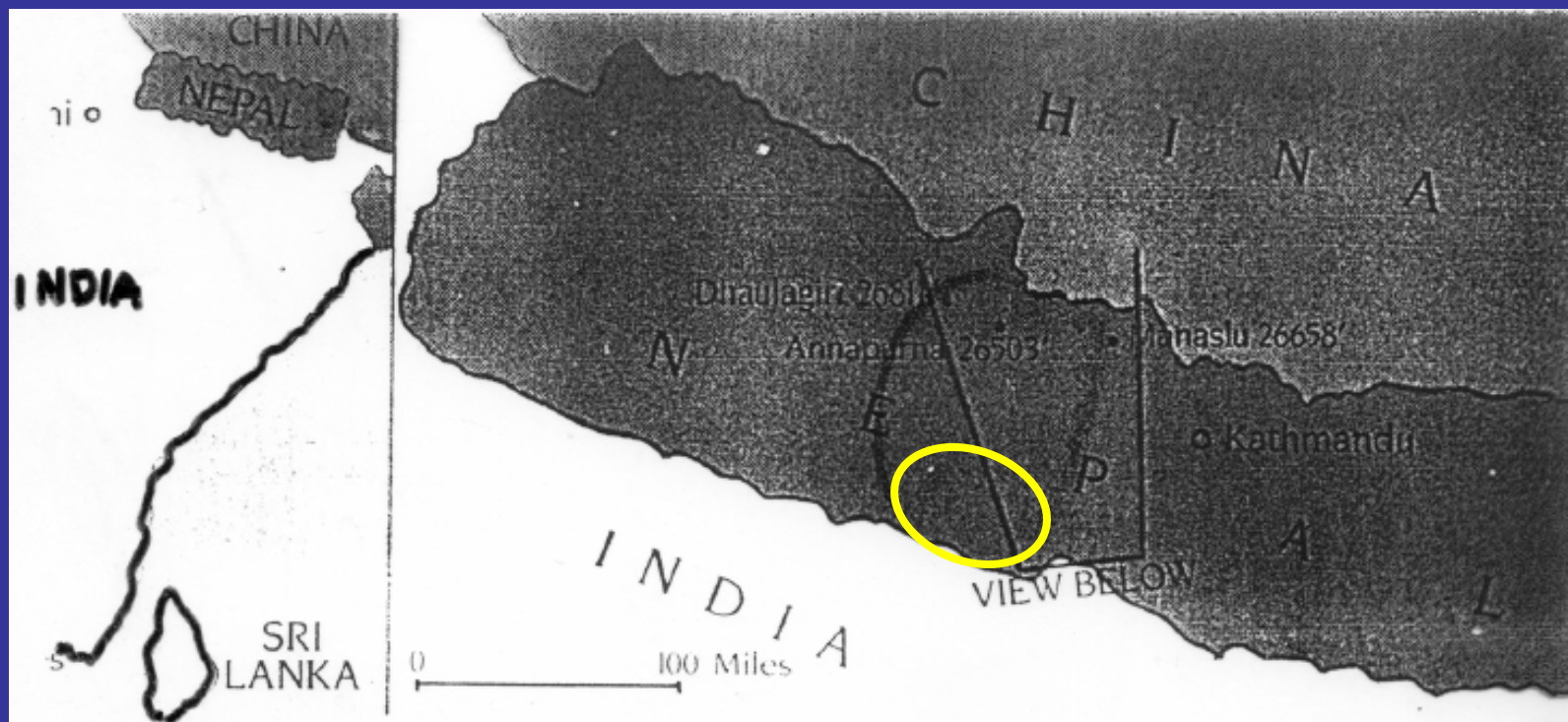
The race is not always to the swift, nor the battle to the strong, for time and chance happens to us all.

Ecclesiastes 9:11



Assumptions of Hardy-Weinberg model

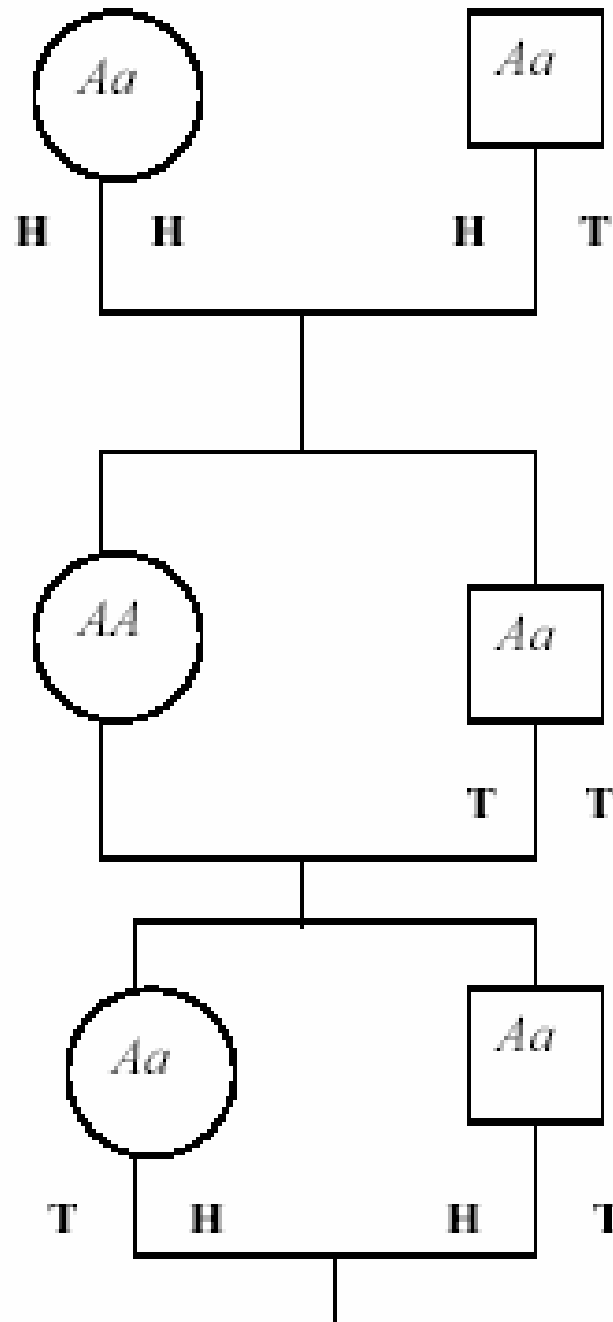
1. Random mating.
2. No mutation.
3. Large (infinite) population size.
4. No differential survival or reproduction
(i.e., no natural selection).
5. No immigration



45 females & 20 males; $N = 65$

Genetic Drift

$$N = 2$$



Work with a student beside to you. One person will be a selfing "plant", and the other will be a "recorder". The plant is initially heterozygous (*HT*) at the coin-flipping locus which has two alleles: *H* & *T*. The plant-person will flip a coin twice to determine the genotype of the next generation. The experiment ends when the plant is homozygous: *HH* or *TT*.

Record the following information:

GENERATION

GENOTYPE

0

HT

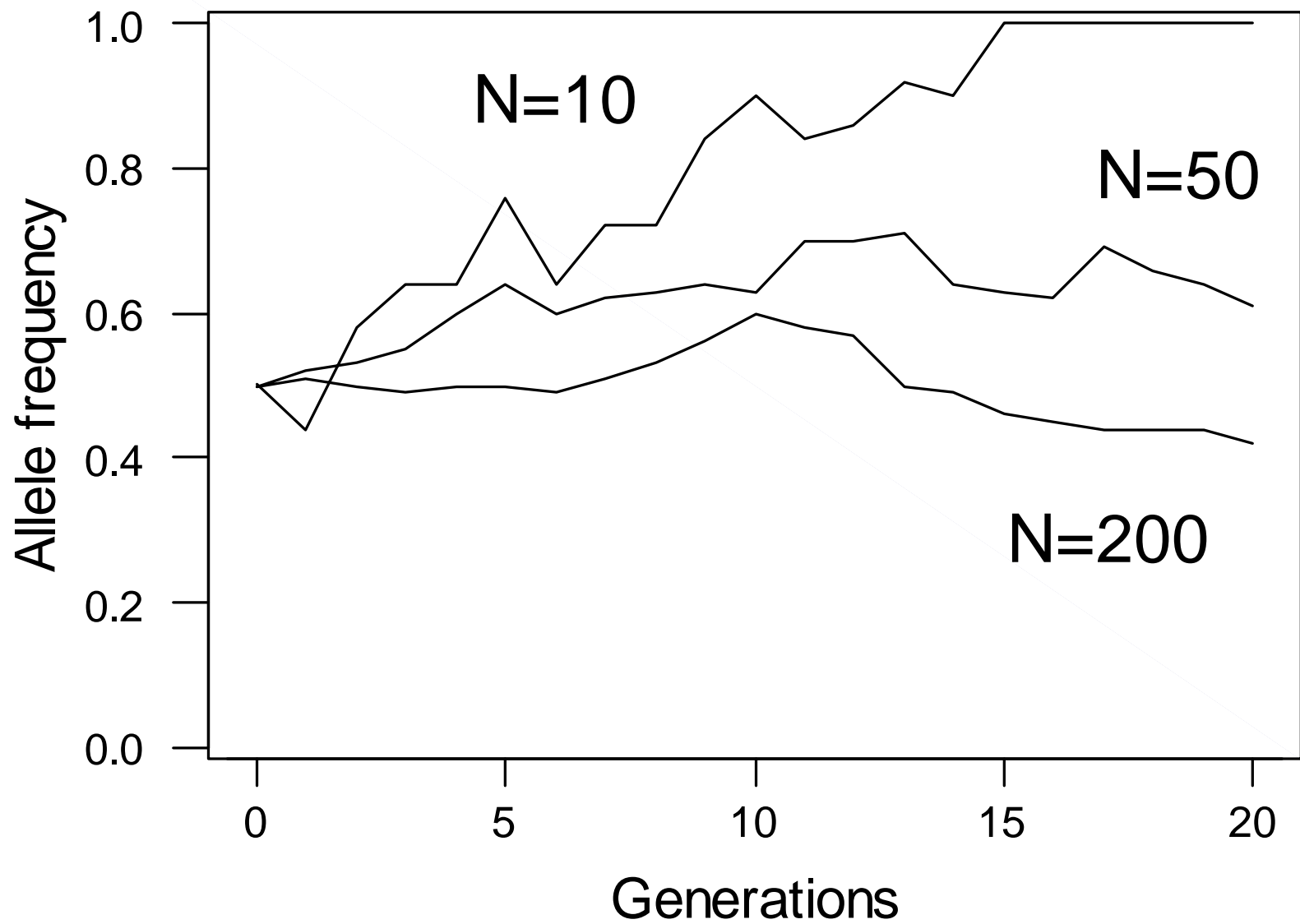
1

2

3

4

Gen	<i>HH</i>	<i>HT</i>	<i>TT</i>	<i>H</i> obs	<i>H</i> * exp
0				1.00	1.00
1					0.50
2					0.25
3					0.12
4					0.06
5					0.03
6					0.02
7					0.01
8					~0
Total				---	---



Genetic drift has two primary effects on the genetic composition of populations:

(1) Change in allele frequencies.

$$p' = p \pm 2\sqrt{\frac{(pq)}{(2N)}}$$

(2) Loss of genetic variation.

$$\Delta h = -\frac{1}{2N}$$

(1) Change in allele frequencies.

$$p' = p \pm 2 \sqrt{\frac{(pq)}{(2N)}}$$

95% interval

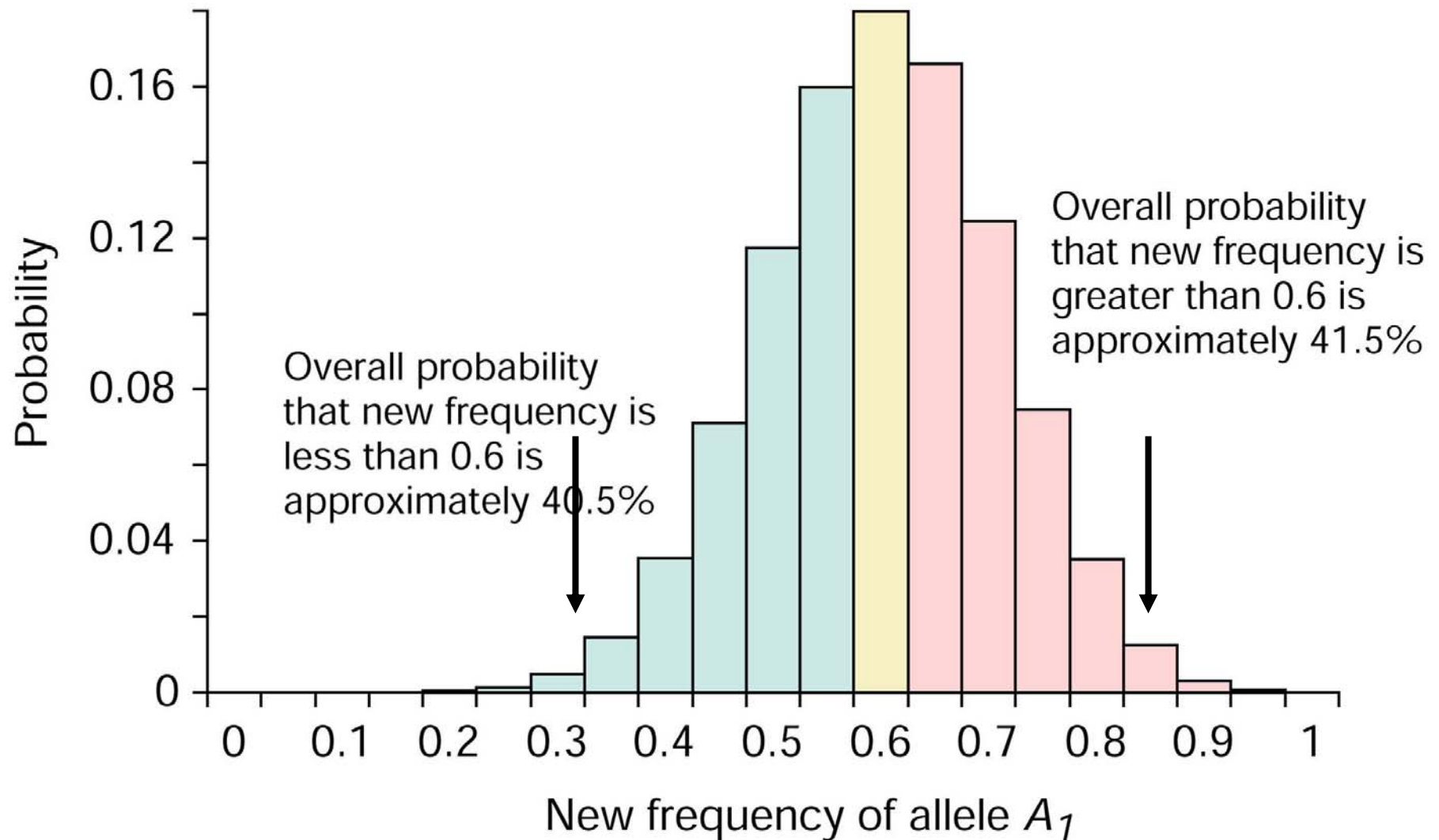
$$p = 0.6; N = 10$$

$$p' = 0.6 \pm 2 (0.11) = 0.6 \pm 0.22$$

$$0.28 - 0.82$$

95% of the time

Probability that new frequency is exactly 0.6 is approximately 18%



Populus

Simulations of Population Biology

Don Alstad

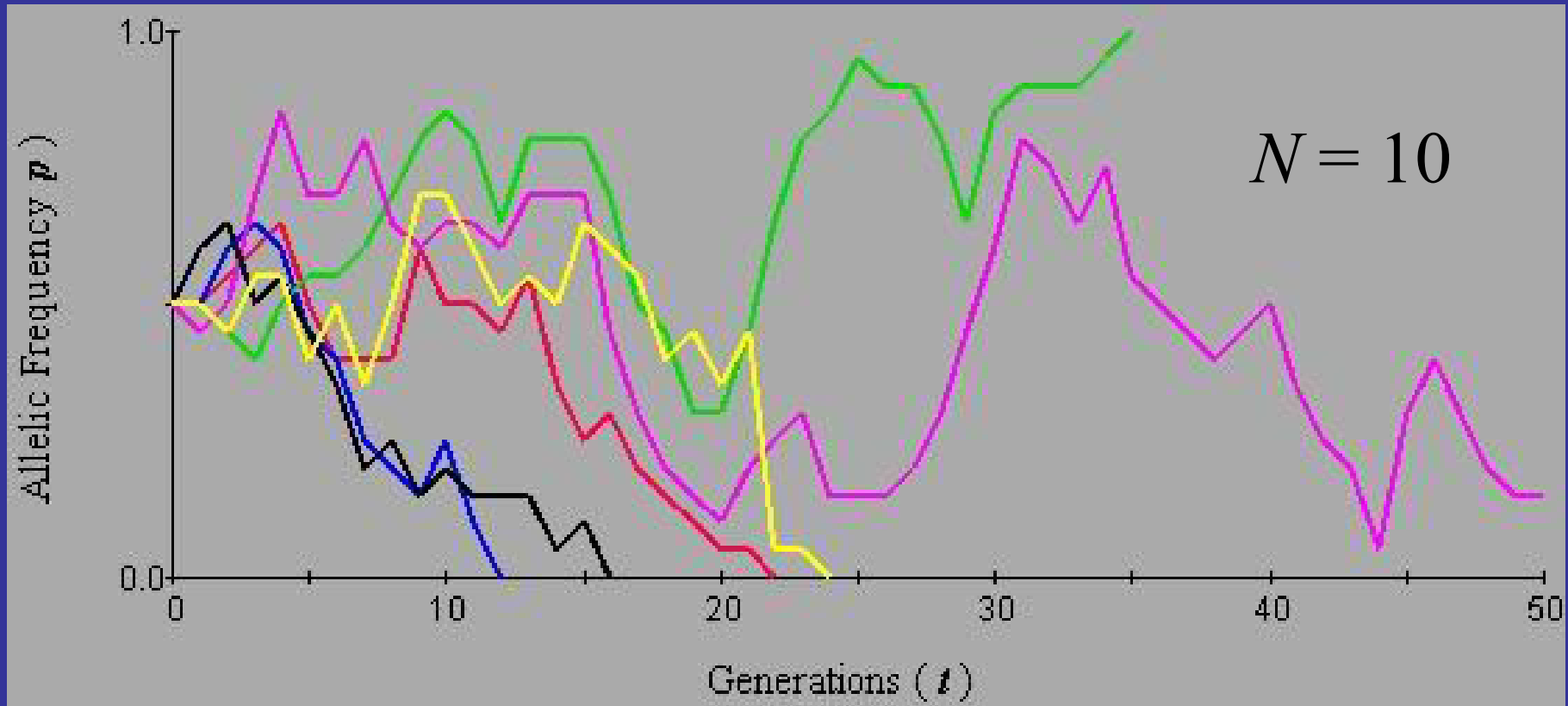
*Department of Ecology, Evolution & Behavior
University of Minnesota*

Programming by

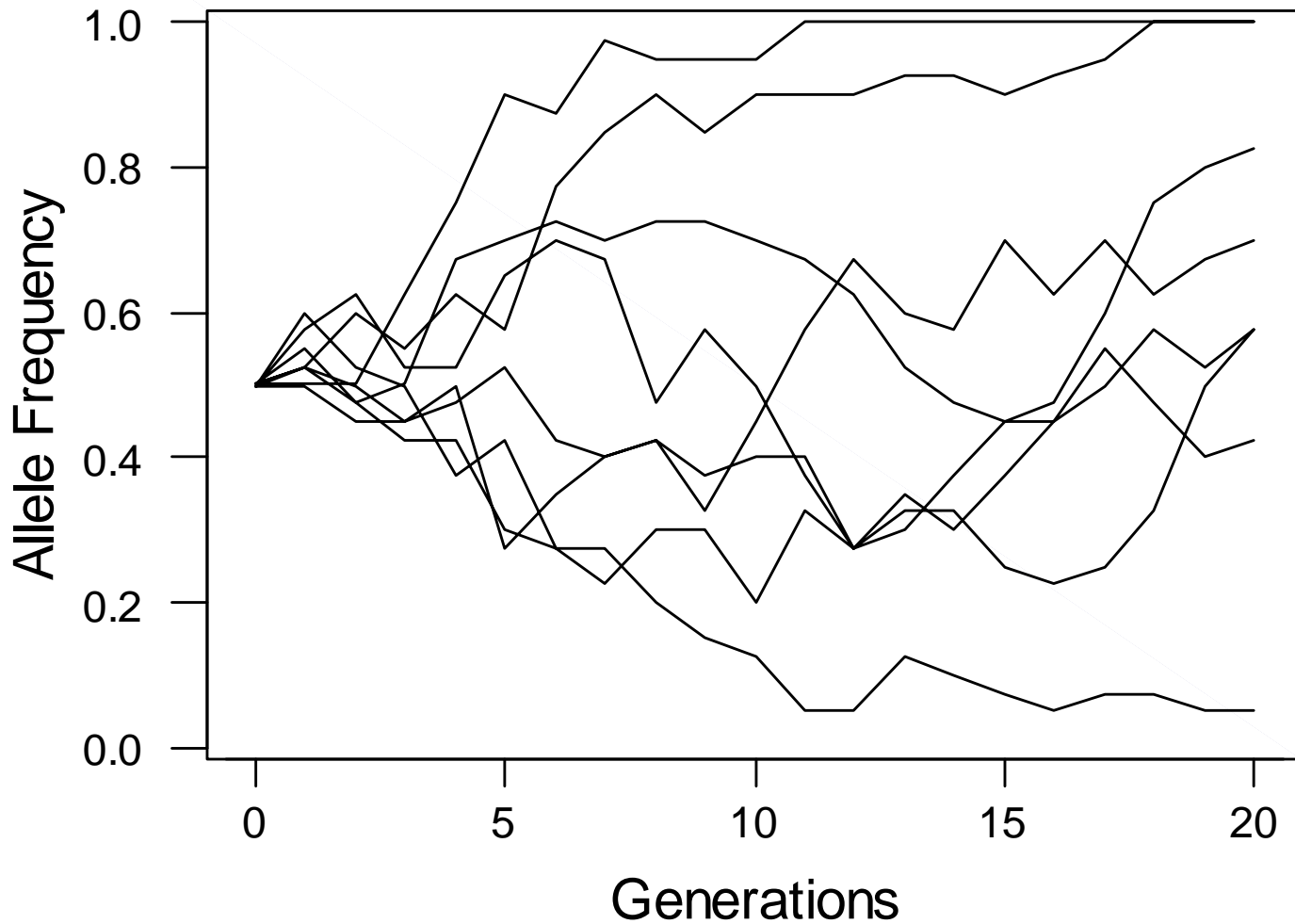
*Amos Anderson, Lars Roe, Sharareh Noorbaloochi
and Chris Bratteli*

Version 5.3

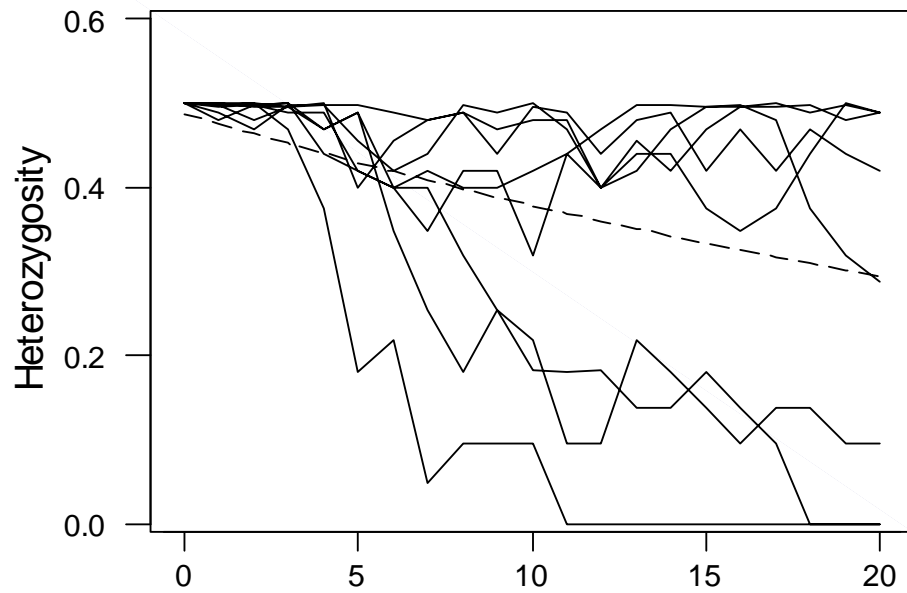
(2) Loss of genetic variation.



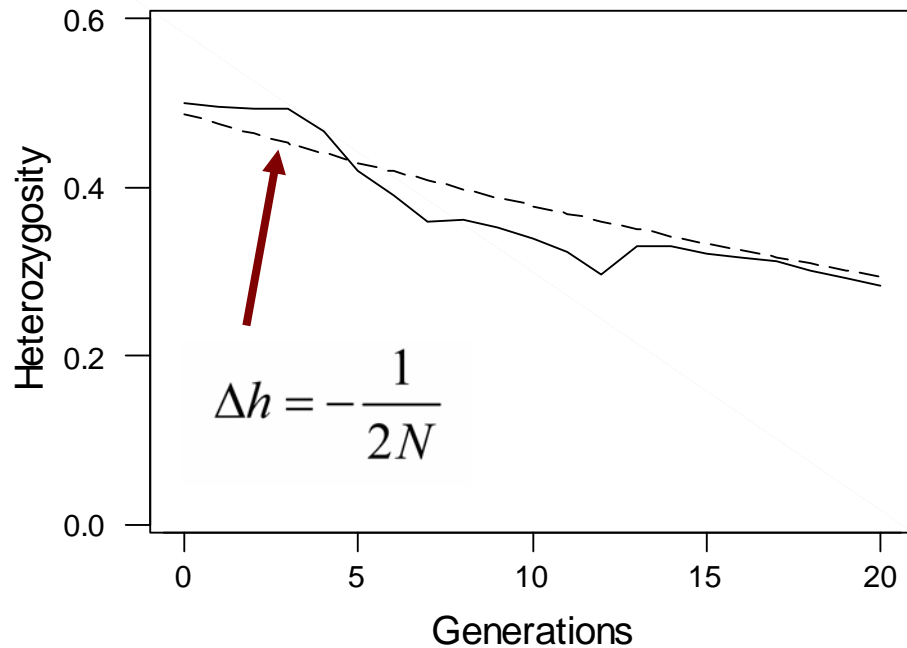
$$\Delta h = -\frac{1}{2N}$$



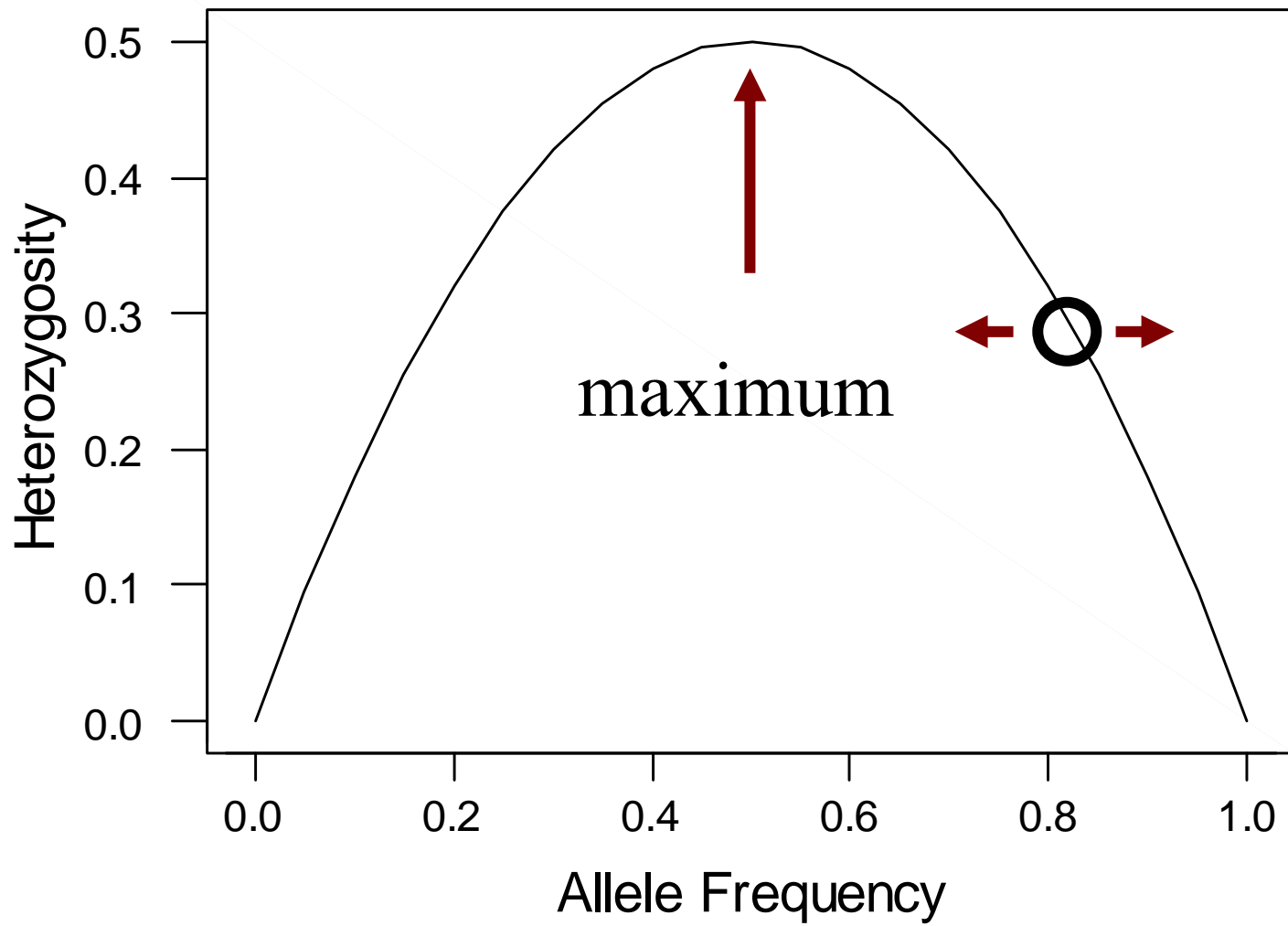
Allele frequency drift at 8 loci



Heterozygosity
at 8 loci



Mean
heterozygosity
at 8 loci



According to

$$\Delta h = -\frac{1}{2N}$$

only 50% of the heterozygosity will be lost during a “bottleneck” of just two individuals.

Do you think that just two individuals will contain 50% of the genetic variation in a population or species?

Why not?

Measures of genetic variation:

- (1) Heterozygosity
- (2) Allelic diversity

Two individuals can carry a maximum of four alleles.

SHORT COMMUNICATION

Analysis of microsatellite DNA from old scale samples of Atlantic salmon *Salmo salar*: a comparison of genetic composition over 60 years

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Abstract

Microsatellite analysis was applied to scale samples of Atlantic salmon collected up to 60 years ago. Samples from the 1930s, from a now endangered Danish population, were compared with recent samples (1989), to test if the present population consists of descendants from the original one. Allele frequencies had changed over time, but individuals from the two samples caught about 60 years apart clustered together when compared with the closest neighbouring population and another reference population. However, fewer alleles were detected in the recent sample from the endangered population, most likely due to a population bottleneck or sampling artefacts.

Locus	Allele	Skjern River 1989	Skjern River 1930s	Conon	Ätran
SSOSL 85	177		0.007		
	179		0.010		
	181		0.007		
	183	0.11	0.316	0.08	0.15
	187		0.003		
	189		0.003	0.46	0.14
	191		0.003	0.02	0.02
	193	0.06	0.076	0.08	0.25
	195	0.40	0.330	0.02	0.02
	197		0.017	0.04	0.13
	199	0.28	0.073	0.21	0.11
	201	0.14	0.142	0.02	0.06
	203			0.08	0.04
	209				0.02
	217		0.003		0.04
	221	0.01	0.007		0.02

Locus	Allele	Skjern River 1989	Skjern River 1930s
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	197		0.017
	199	0.28	0.073
	201	0.14	0.142
	203		
	209		
	217		0.003
	221	0.01	0.007

H_E 0.452 0.528

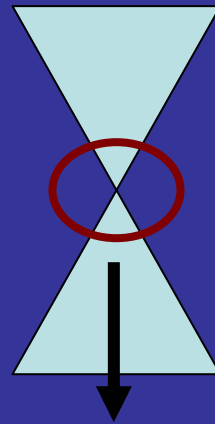
hets + homos = total

$$\frac{n(n-1)}{2} + n = \frac{n(n+1)}{2}$$

$$H_E = 1 - \sum_{i=1}^n p_i^2$$

Population Bottlenecks

Source



Bottleneck

Established

Bottlenecks & Loss of Allelic Diversity

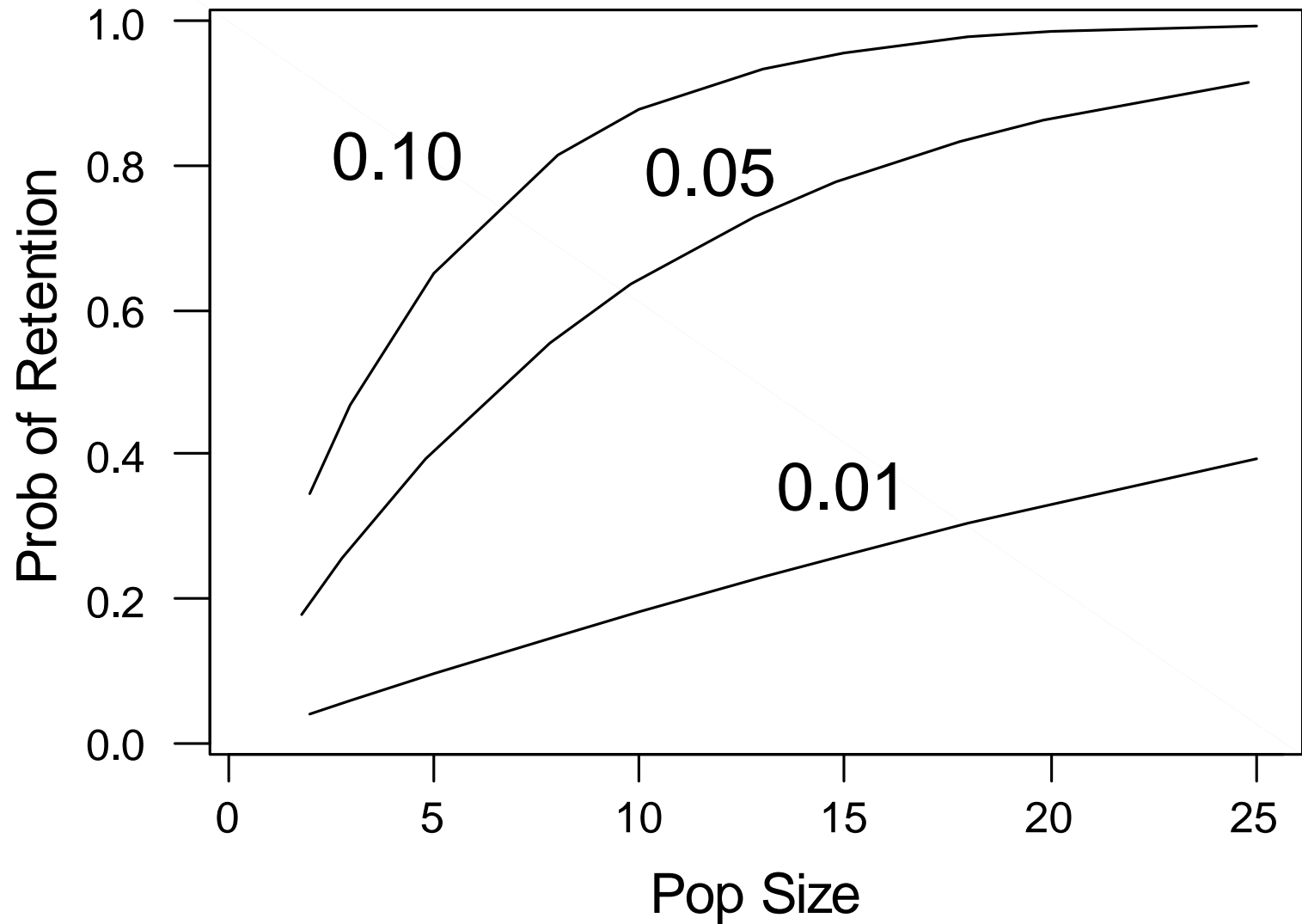
The probability of an allele being lost during a bottleneck of size N is

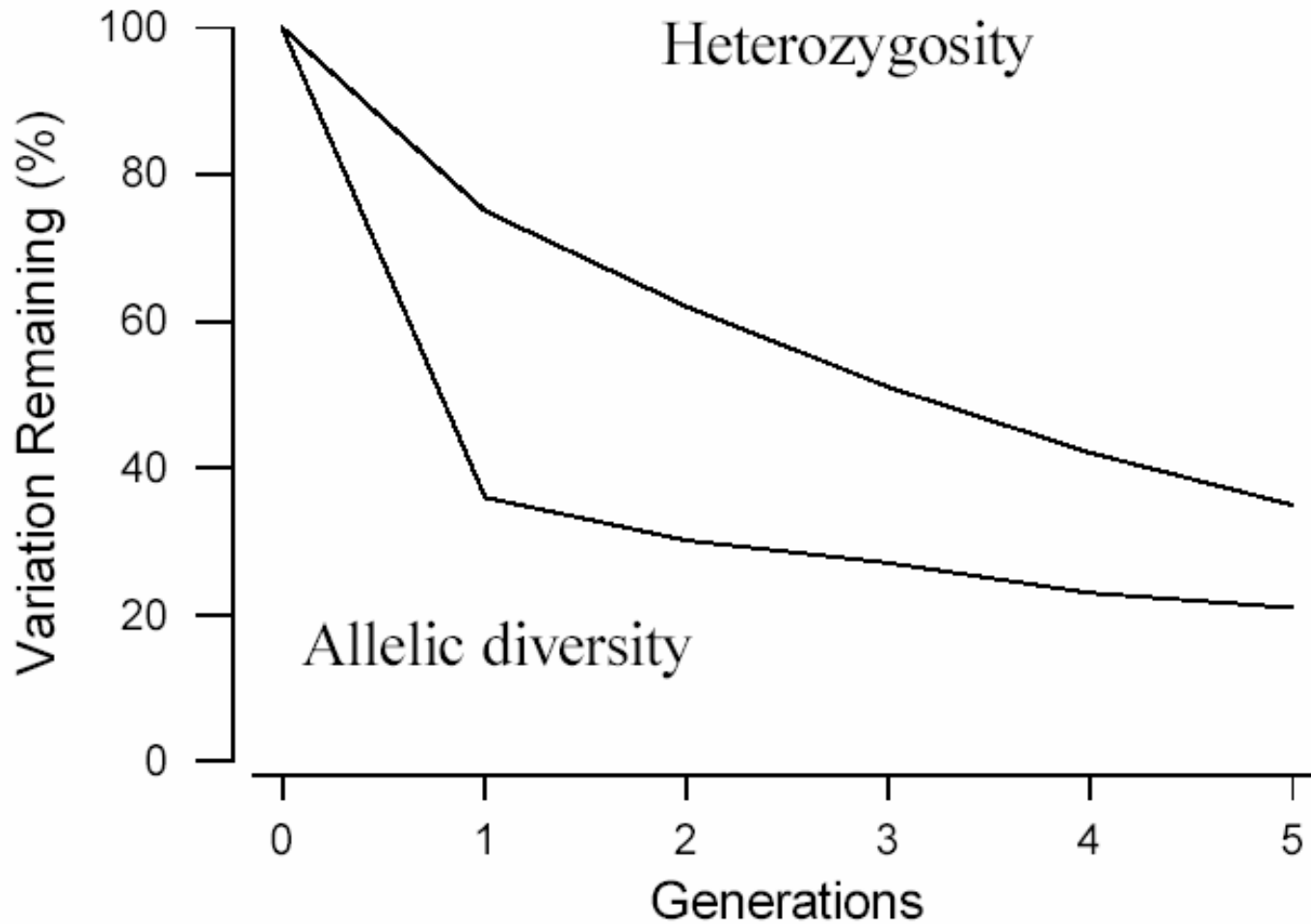
$$(1-p)^{2N}$$

In general, if a population is reduced to N individuals for one generation then the expected total number of alleles (n') remaining is

$$E(n') = n - \sum_{j=1}^n (1 - p_j)^{2N}$$

A bottleneck will cause loss of rare alleles.





Brown bears
(8 microsatellite loci)

Allelic diversity is generally lost more quickly than heterozygosity.

Effects of genetic drift

(1) Changes in allele frequency

Increase in frequency of harmful alleles.

(2) Loss of allelic diversity

For example, MHC & disease resistance

(3) Inbreeding and loss of heterozygosity

Inbreeding effect of small populations

(1) Changes in allele frequency

Increase in frequency of harmful alleles

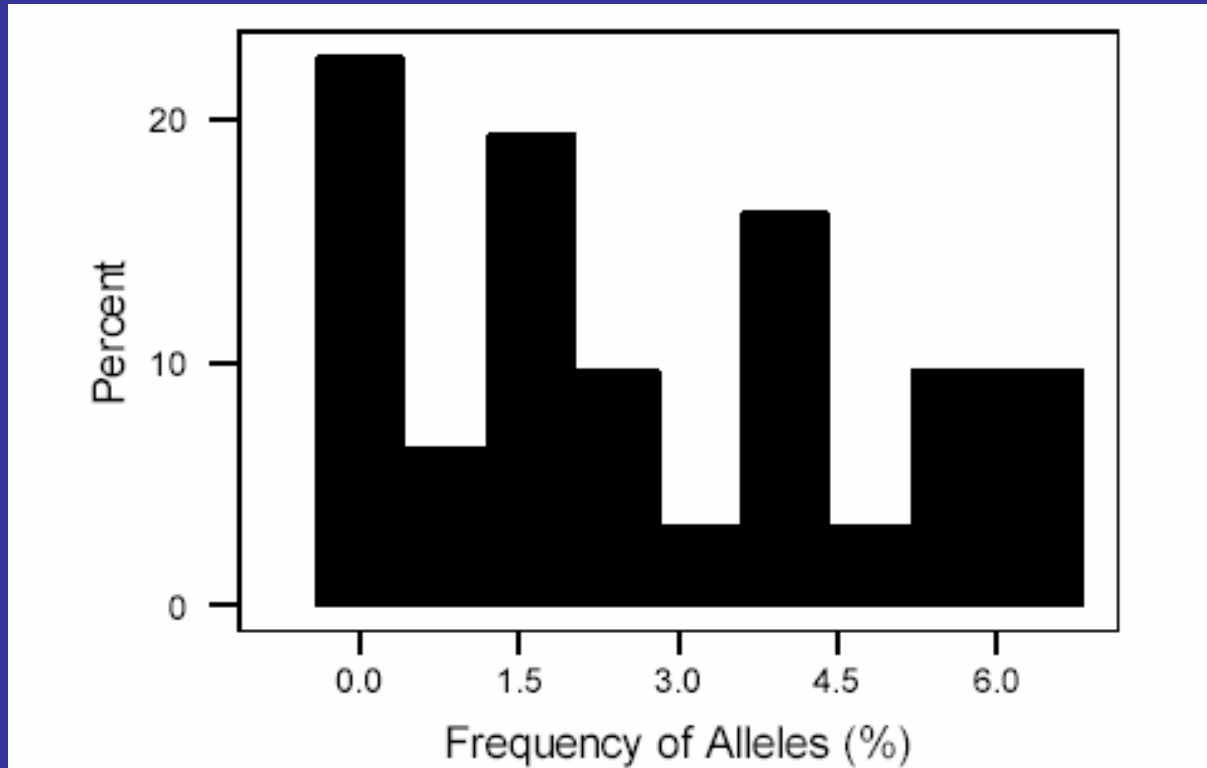


DOG BREED	COMMON PROBLEM
dalmatians	kidney stones; deafness
Afghan hounds	limb paralysis
laplands	muscular dystrophy
beagles	hemolytic anemia
malamutes	anemic dwarfism
golden retrievers	hip dysplasia; lymphatic cancer
Labrador retrievers	dwarfism
collies	blindness
cocker spaniels	hernias

(2) Loss of allelic diversity

For example, MHC & disease resistance

Allelic diversity is much more sensitive to bottlenecks than heterozygosity.



Blackbirds:
37 alleles!

Major Histocompatibility Complex (MHC)

Bottleneck $N=2$

Heterozygosity

$$h = 1 - (1/2N) = 75\% \text{ retained}$$

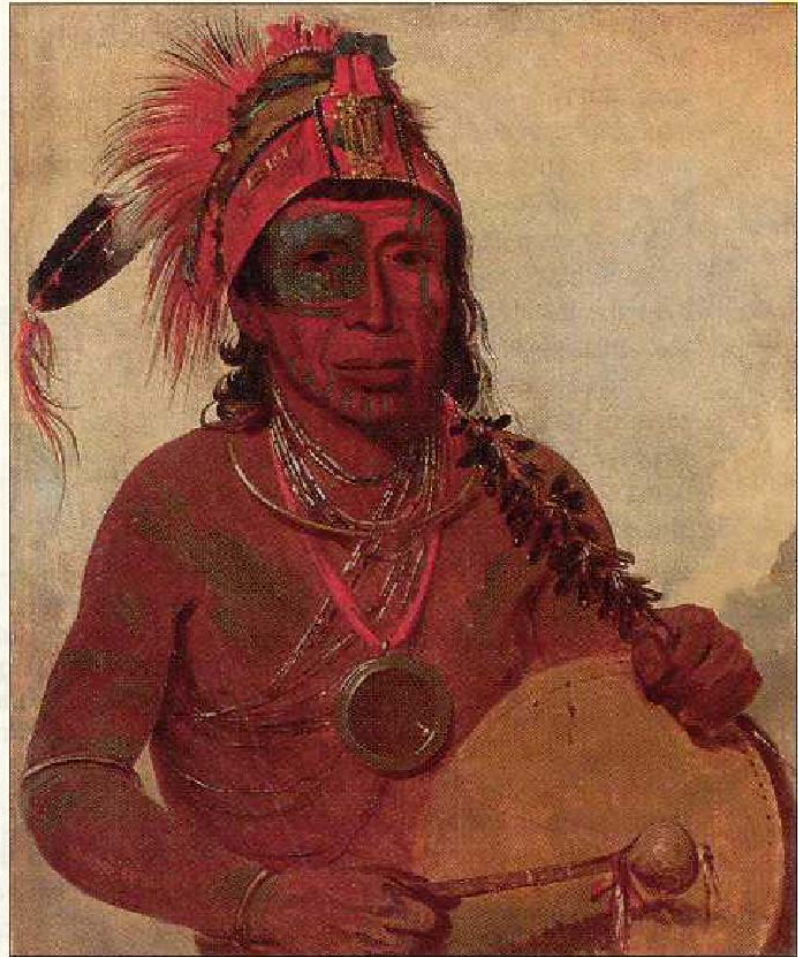
At most 4 of 37 alleles retained:

$$A = 1 - (33/37) = 11\% \text{ retained}$$

Why Did They Die?

Science 258:1739-1740.

1992.



NATIONAL MUSEUM OF AMERICAN ART, WASHINGTON, DC/ART RESOURCE

Blue Medicine, a medicine man of the Ting-ta-to-ah Band (Eastern Sioux). The genetic homogeneity of Amerinds rendered them particularly susceptible to diseases introduced by explorers and immigrants from the Old World. [Painting by George Catlin]

Human MHC alleles (A & B loci).

Human Group	Sample	No. alleles
Africans	1,432	40
Europeans	1,069	37
South Amerinds	1,944	10
North Amerinds	1,163	17
Polynesians	12,243	14

(3) The inbreeding effect of small populations

Inbreeding will occur in small populations and cause a loss of heterozygosity (h).

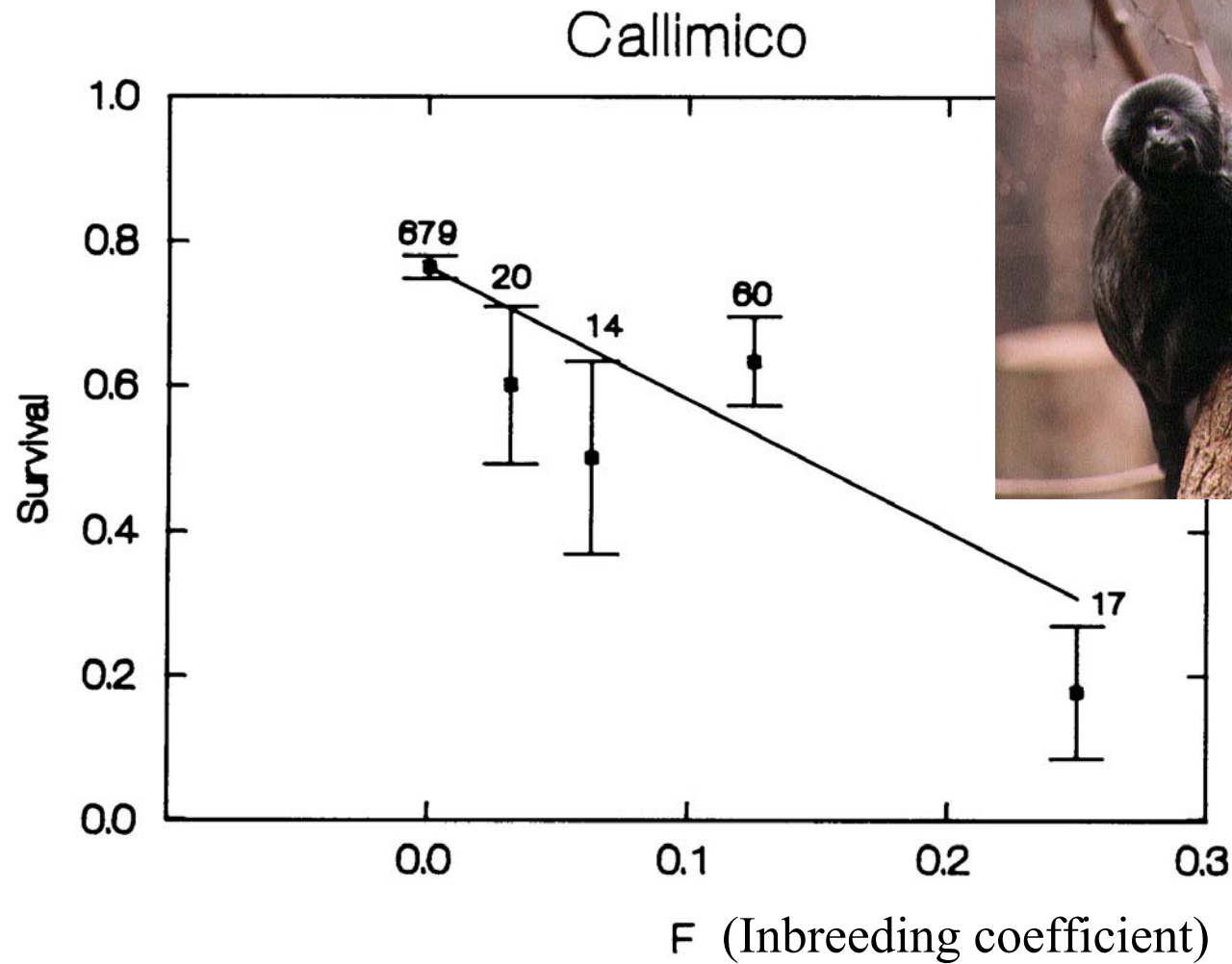
F is the inbreeding coefficient and represents the loss in heterozygosity (h) due to mating between relatives (i.e., $F = 1 - h$). The increase in homozygosity (and an equal loss of heterozygosity) due to genetic drift will occur at the following rate per generation:

$$\Delta F = 1/(2N)$$

$$\Delta h = -1/(2N)$$

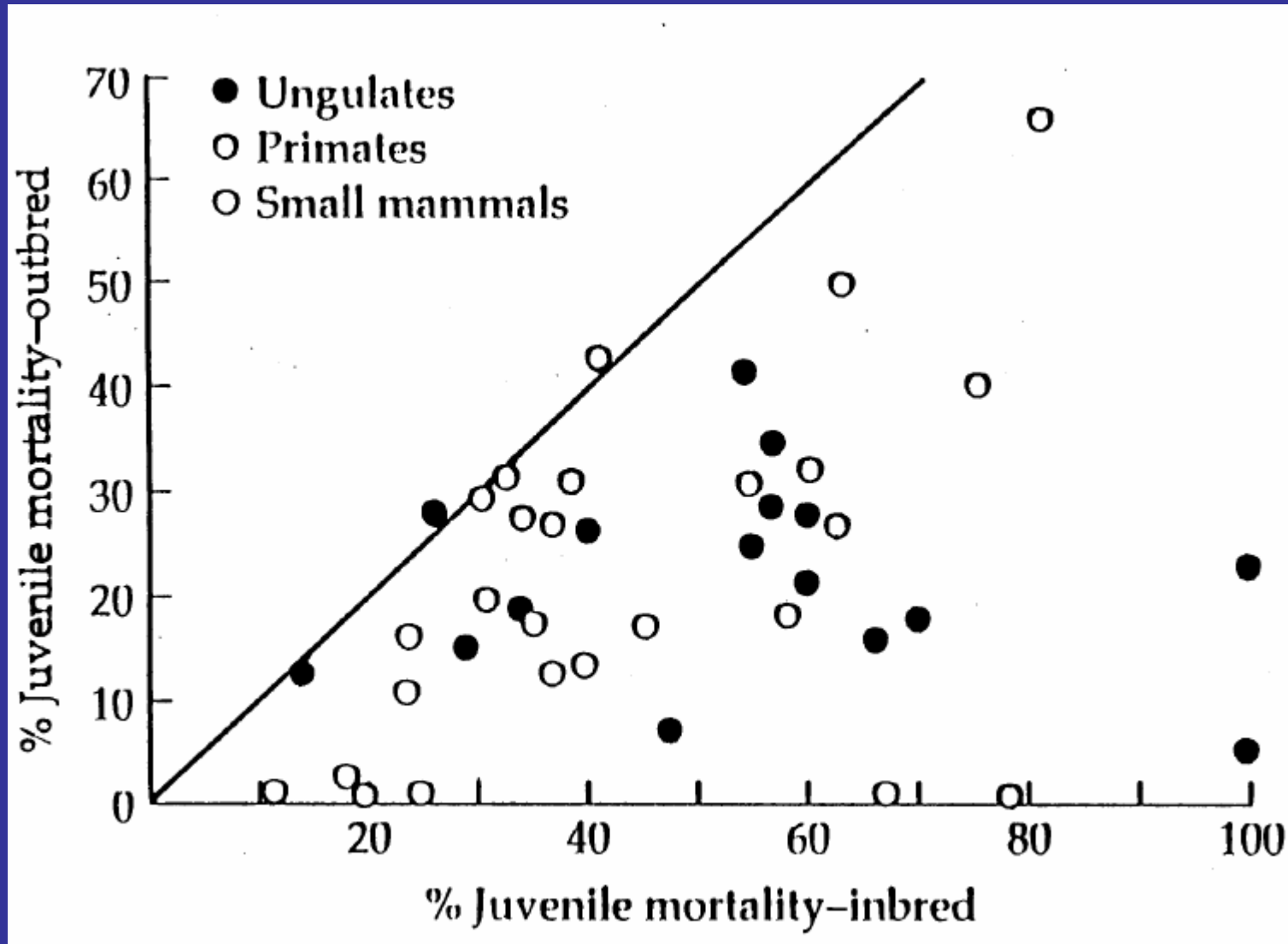
Effects of inbreeding and small population size

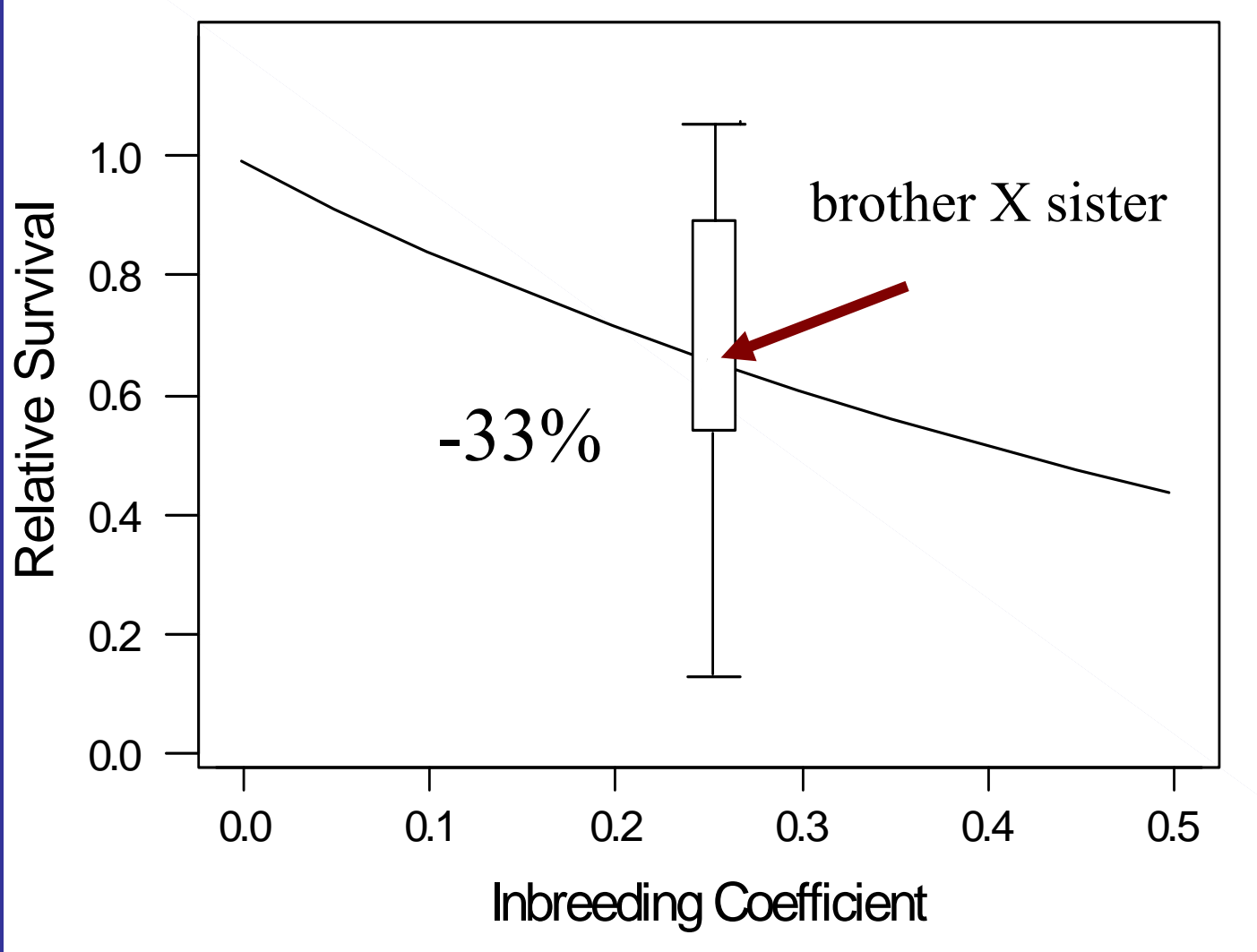
STAGE OF MORTALITY	RATE OF MORTALITY		RELATIVE SURVIVAL
	PARENTS UNRELATED	PARENTS FIRST-COUSINS	
Miscarriage	0.129	0.145	97%
Stillbirths	0.044	0.111	93%
Postnatal	0.024	0.081	94%
Infant	0.089	0.156	93%
Juvenile	0.160	0.229	92%
Young adult	0.031	0.050	98%
	-----	-----	---
Total	0.397	0.568	72%



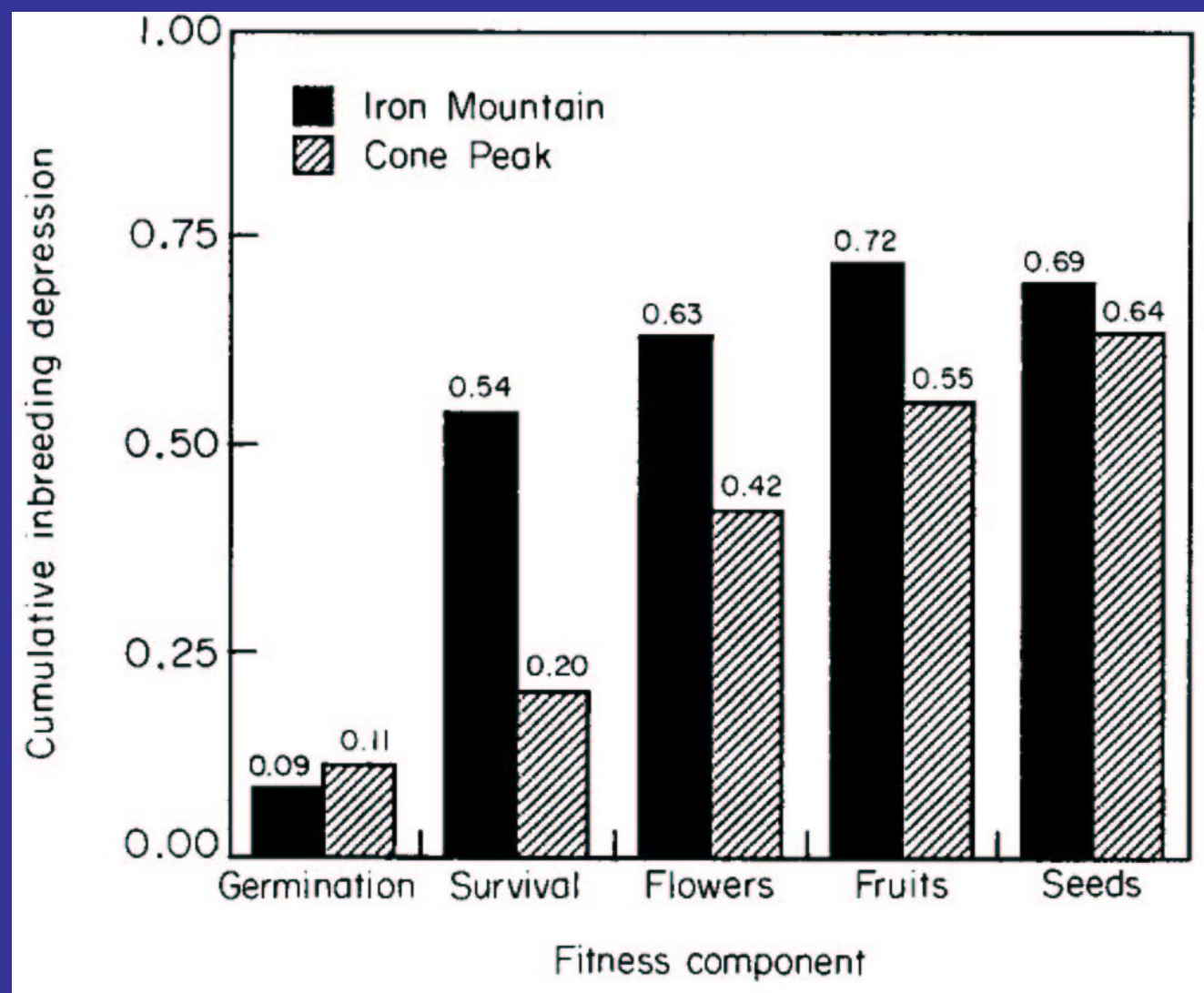
Relationship between inbreeding (F) and infant survival in captive callimico monkeys.

Inbreeding depression in zoo animals





Mean reduction in relative juvenile survival to six months of age in 40 captive mammal populations (Ralls et al. 1988).

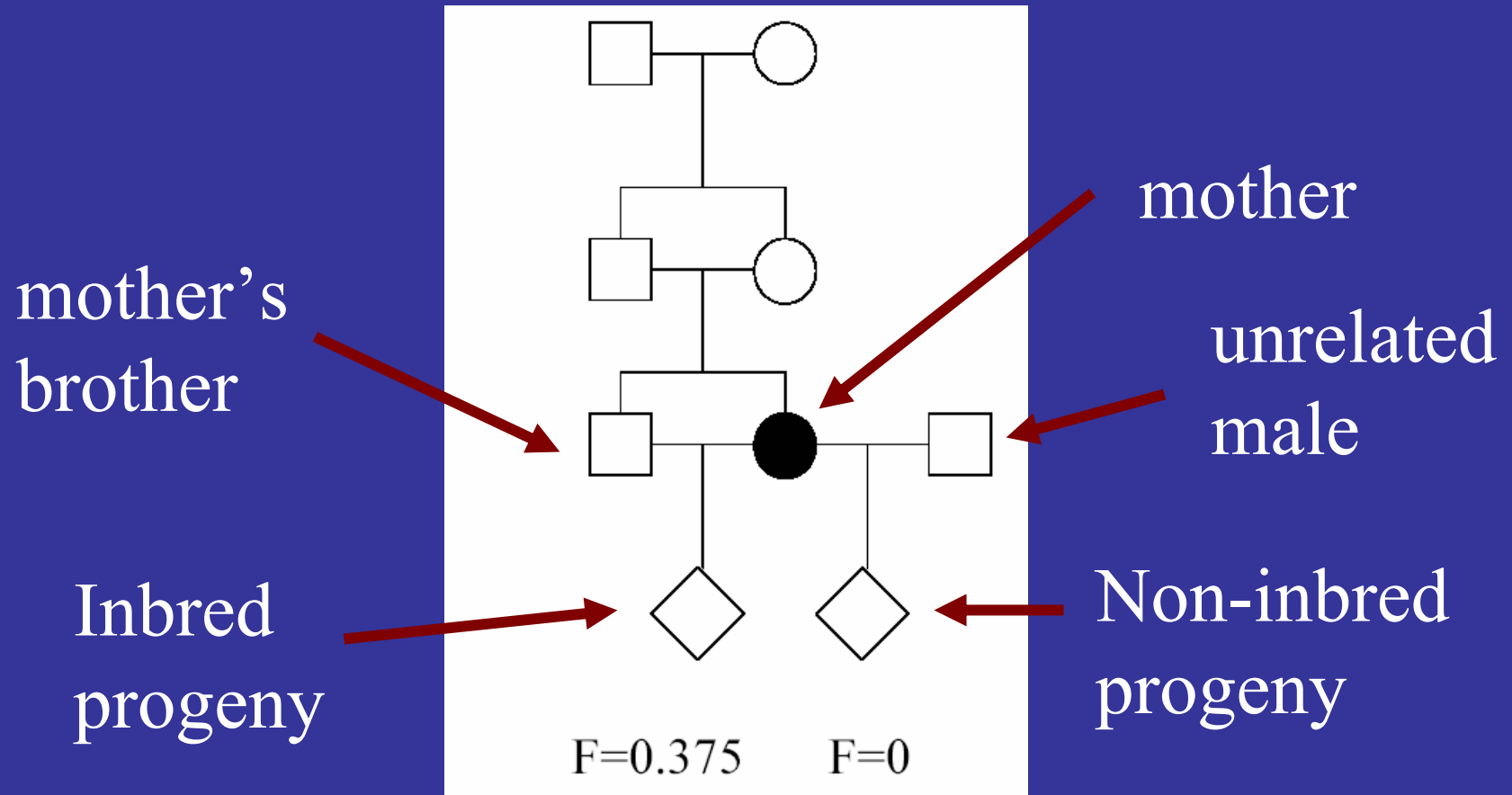


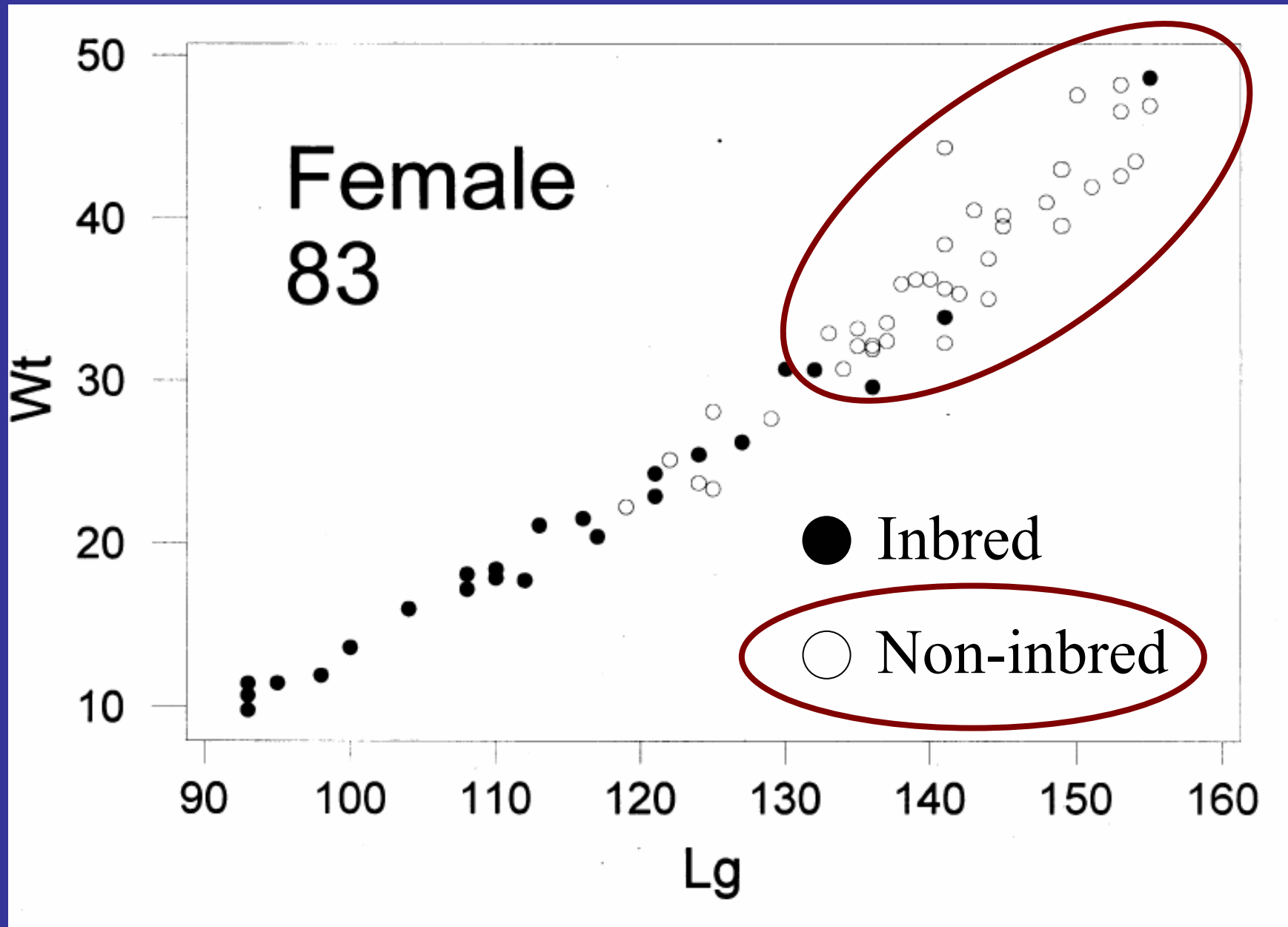
Large reduction in survival of progeny from selfing compared to outcrossing.

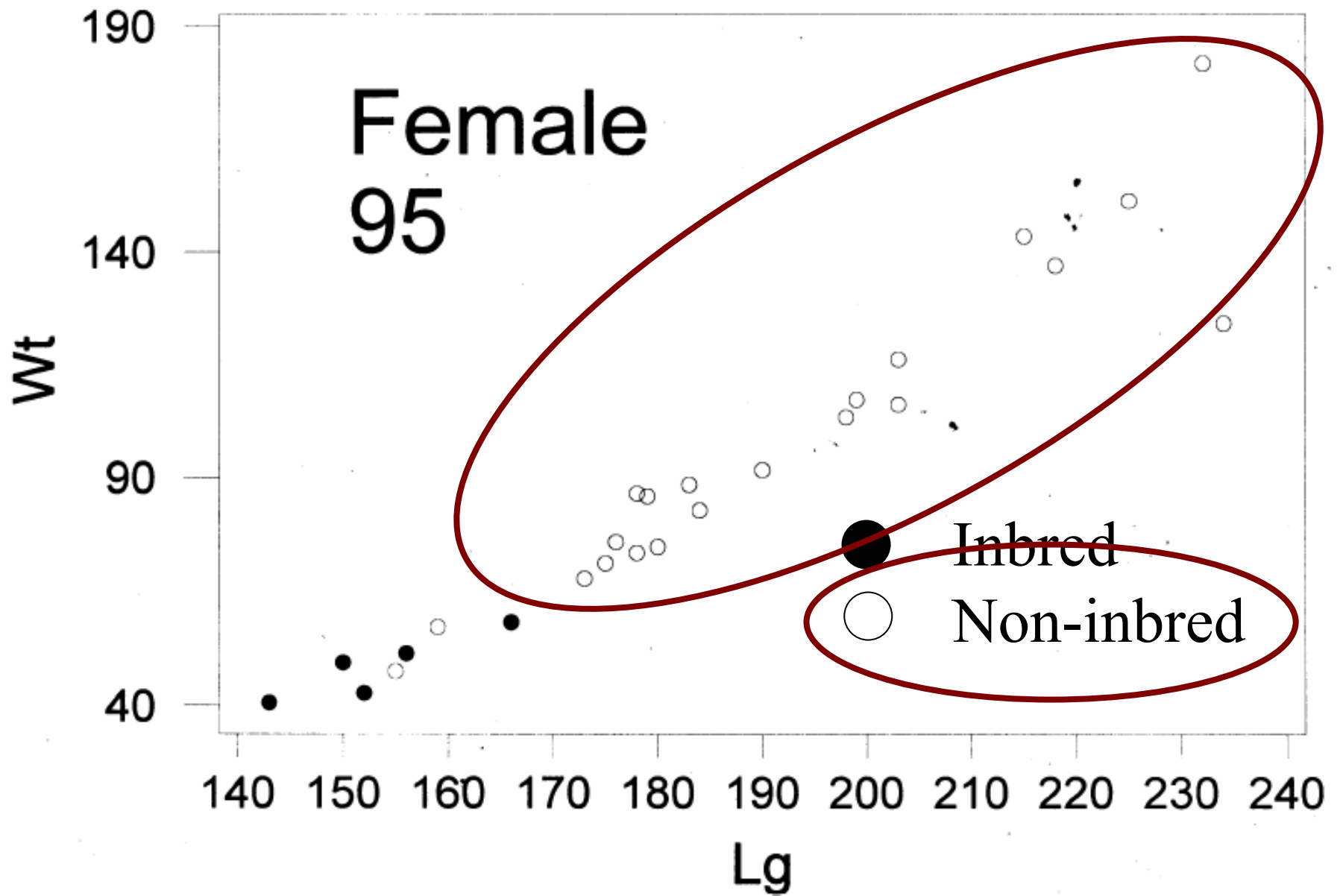


Inbreeding depression due to self fertilization in monkey flowers

Test for inbreeding depression in hatchery rainbow trout.







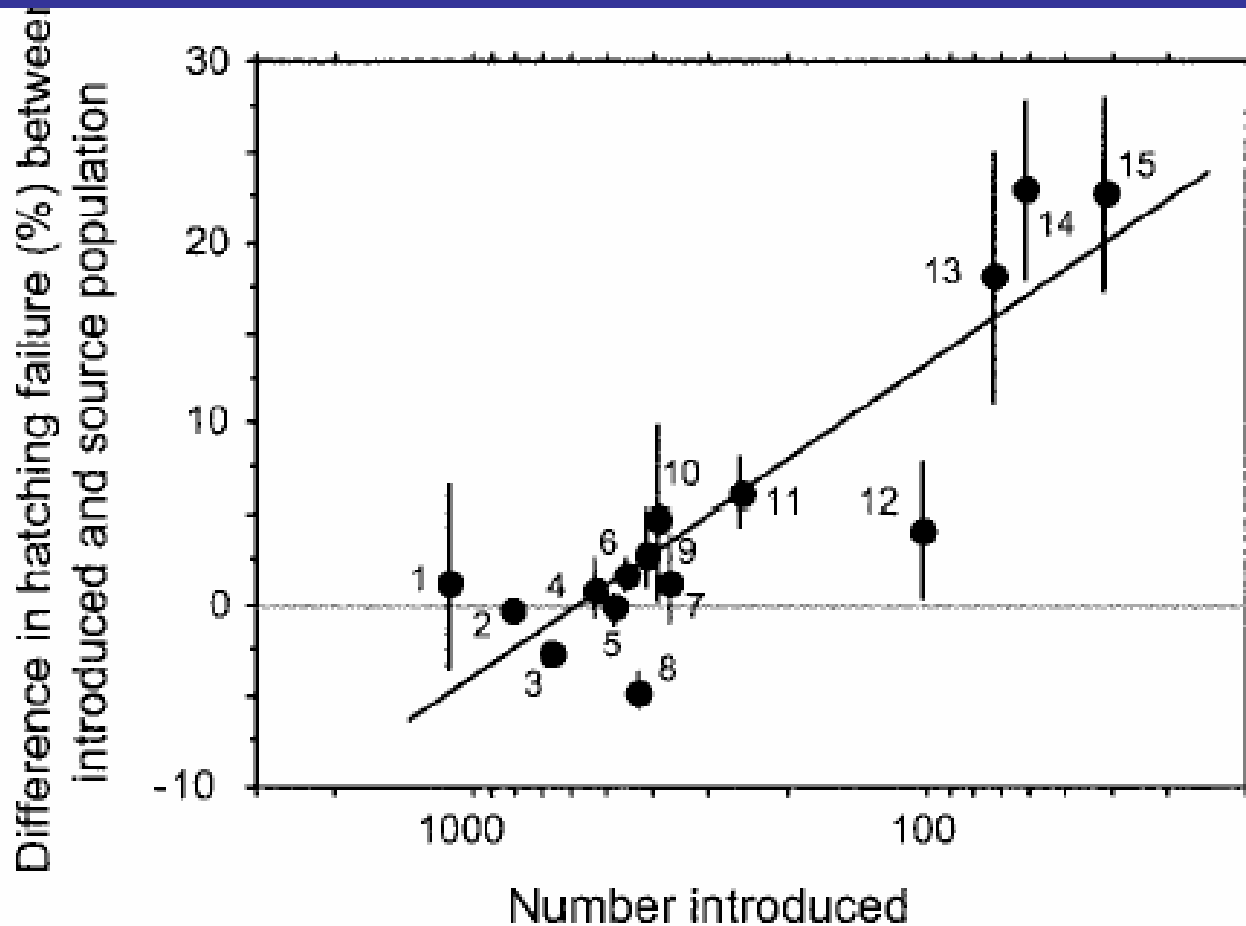
Female		Weight	Length	Survival
83	in	22.57***	116.87***	0.58**
	out	33.85	136.02	1.00
85	in	16.47***	105.50***	0.63*
	out	25.28	123.92	1.00
93	in	25.25*	123.00*	0.08***
	out	39.38	141.08	1.00
95	in	48.49***	153.40***	0.24***
	out	98.70	192.24	1.00
97	in	26.05*	125.70	0.62*
	out	29.22	130.10	1.00
133	in	36.40***	139.33***	0.90
	out	51.42	158.85	1.00
137	in	23.64	119.16	0.83
	out	27.35	126.56	1.00
141	in	20.5***	109.1***	0.71
	out	35.0	128.1	1.00

Mean
-43%

Hatching failure increases with severity of population bottlenecks in birds

James V. Briskie* and Myles Mackintosh

School of Biological Sciences, University of Canterbury, Private Bag 4800, Christchurch, New Zealand



(15) rook
(14) Canada goose
(13) myna

(12) greenfinch

CHAPTER 7

EFFECTIVE POPULATION SIZE

Effective population size (N_e) is one of the most fundamental evolutionary parameters of biological systems, and it affects many processes that are relevant to biological conservation.



EFFECTIVE POPULATION SIZE (N_e) is the size of the ideal population (N) that will result in the same amount of genetic drift as in the actual population being considered.

$$\Delta h = -1/2N$$

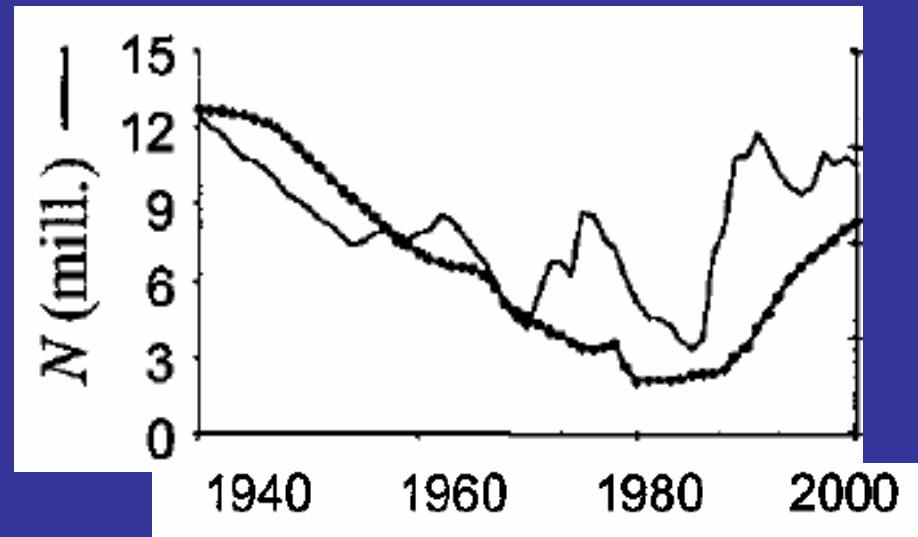
Effective population size is whatever must be substituted in the formula ($1/2N$) to describe the actual loss in heterozygosity.

Sewall Wright (1969)

Loss of microsatellite diversity and low effective population size in an overexploited population of New Zealand snapper (*Pagrus auratus*)

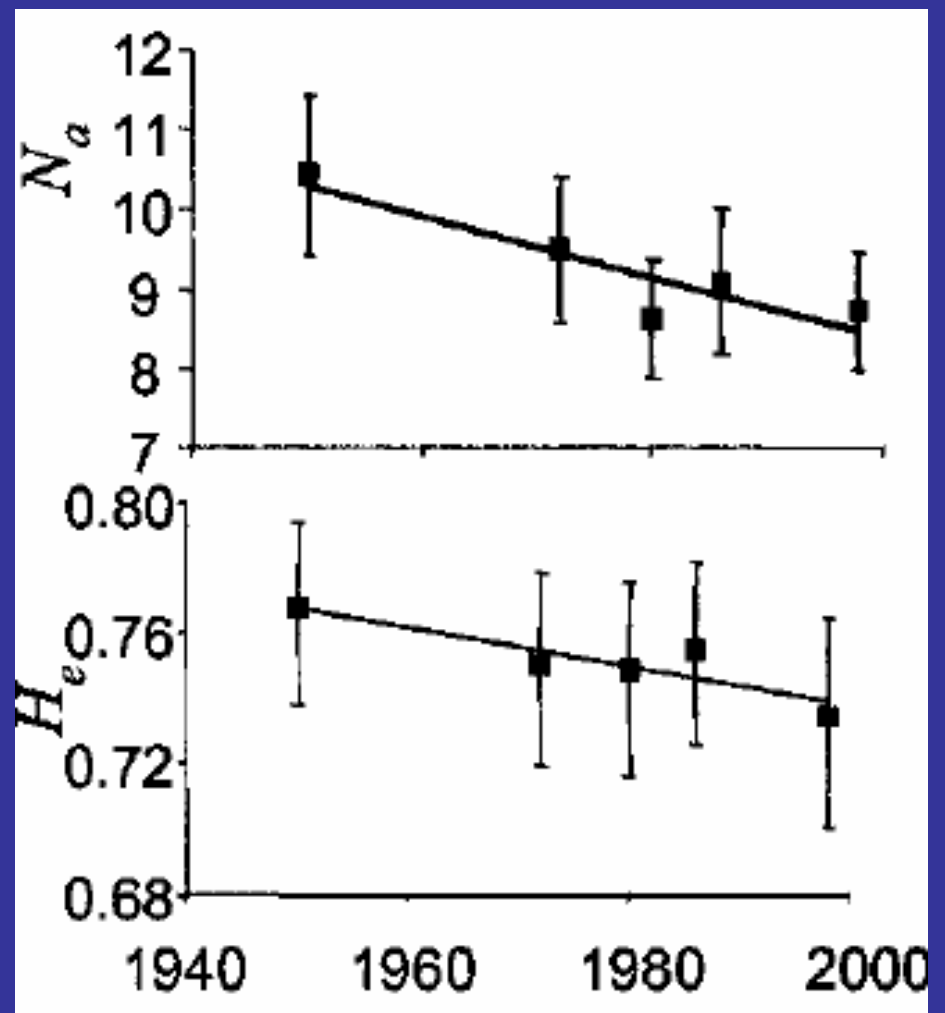
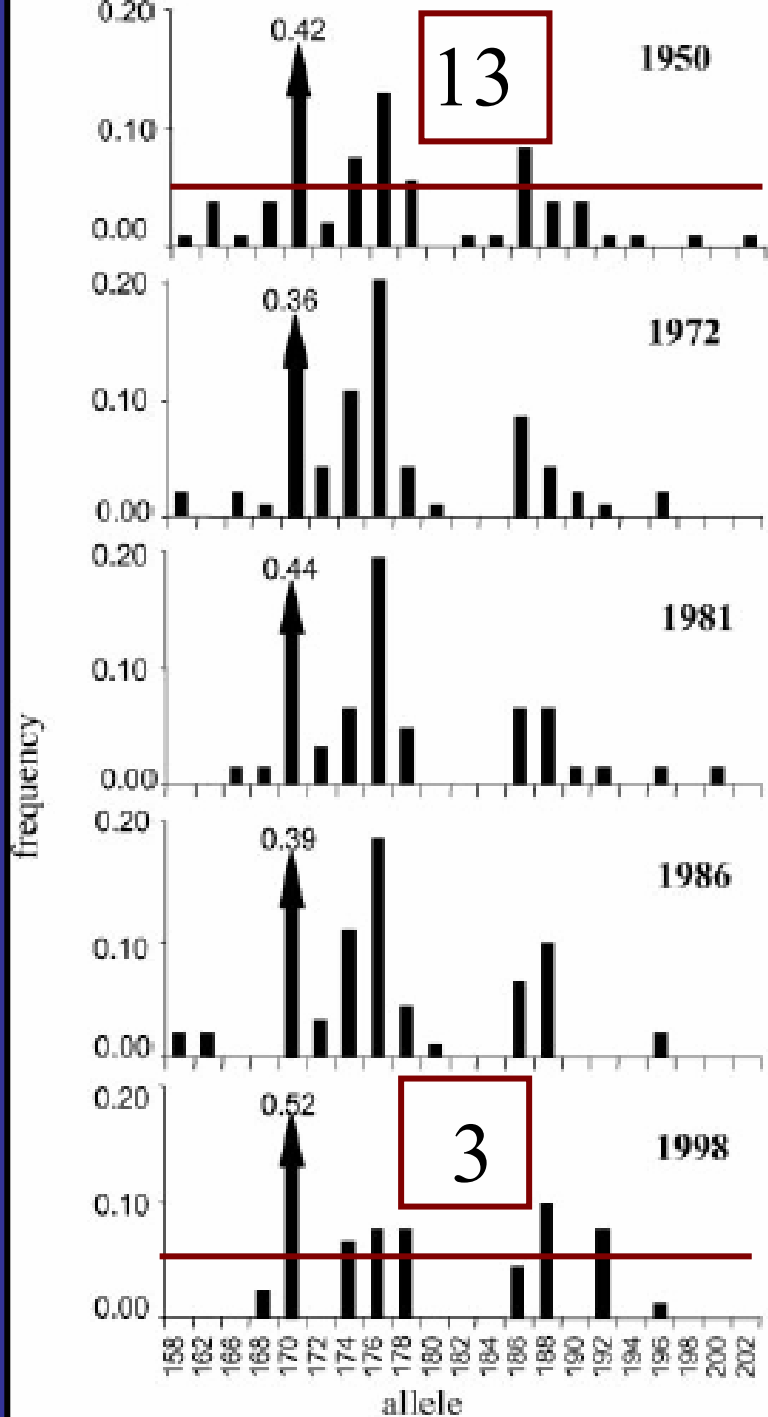
Lorenz Hauser^{*†}, Greg J. Adcock^{*†}, Peter J. Smith[‡], Julio H. Bernal Ramírez^{*¶}, and Gary R. Carvalho^{*}

PNAS 99:11742-11746 (2002)



Orange roughy

Census Pop Size (N_C)



$N_e = 180 (80 - 720, 95\% \text{ CI})$

$$\Delta h = -1/2N$$

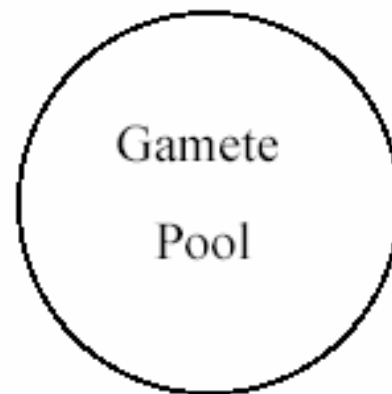
Why?

$$h = 1.0$$

$$N = 10$$

$a1a2$	$a3a4$	$a5a6$
$a7a8$	$a9a10$	$a11a12$
$a13a14$	$a15a16$	$a17a18$
	$a19a20$	

Equal contribution
of 10 individuals



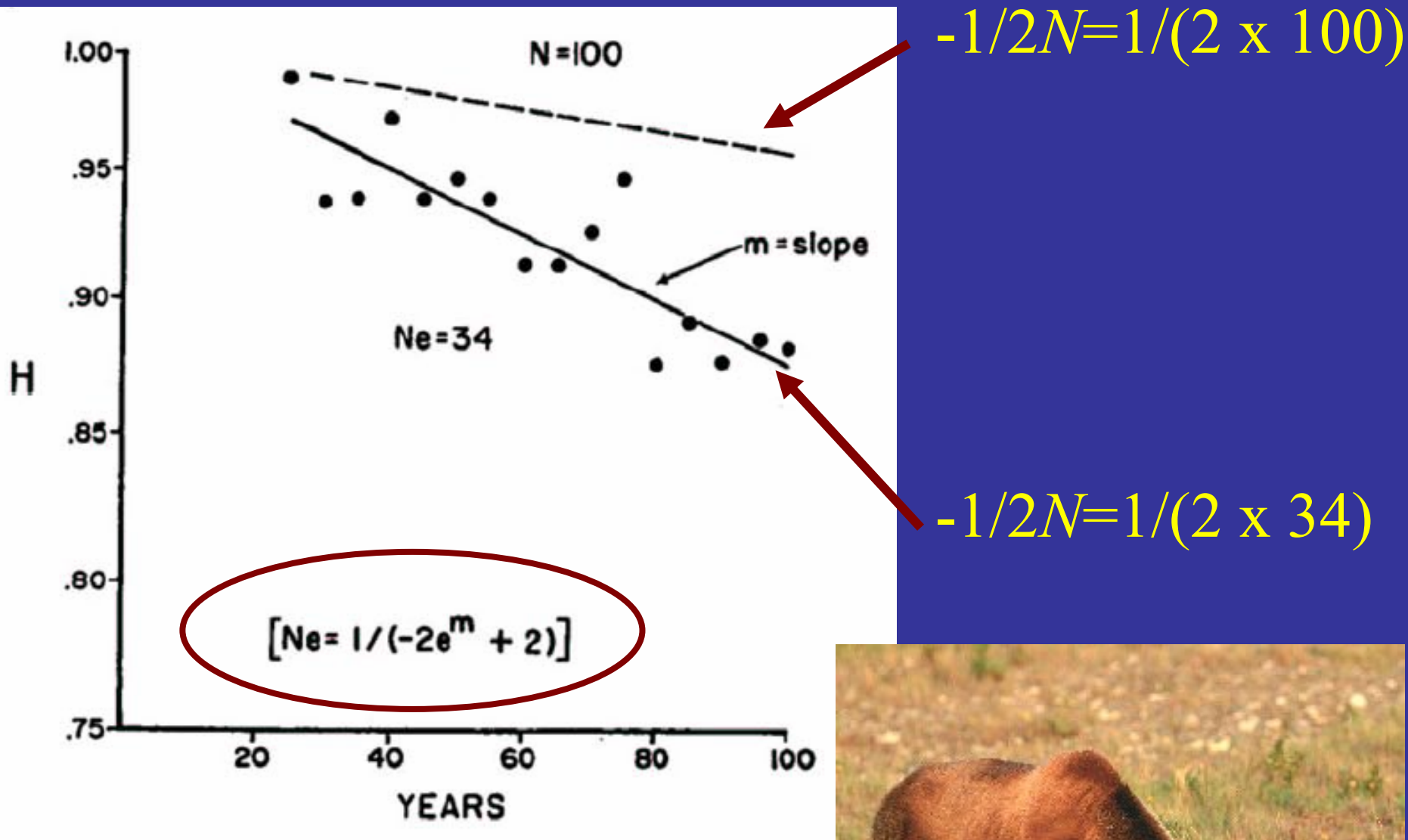
Equal frequency
of 20 alleles

Randomly pick
two gametes

$$h = 1 - (1/2N) = 0.95$$

Assumptions of Ideal Population

- (1) Equal numbers of males and females.
- (2) All individuals have an equal probability of contributing an offspring to the next generation.
- (3) Constant population size.
- (4) Non-overlapping (discrete) generations.



Harris, R. B., & F. W. Allendorf. 1989.
Genetically effective population size of large
mammals an assessment of estimators.
Conservation Biology 3:181-191.



Unequal Sex Ratio

$$\frac{1}{N_e} = \frac{1}{4 N_f} + \frac{1}{4 N_m}$$

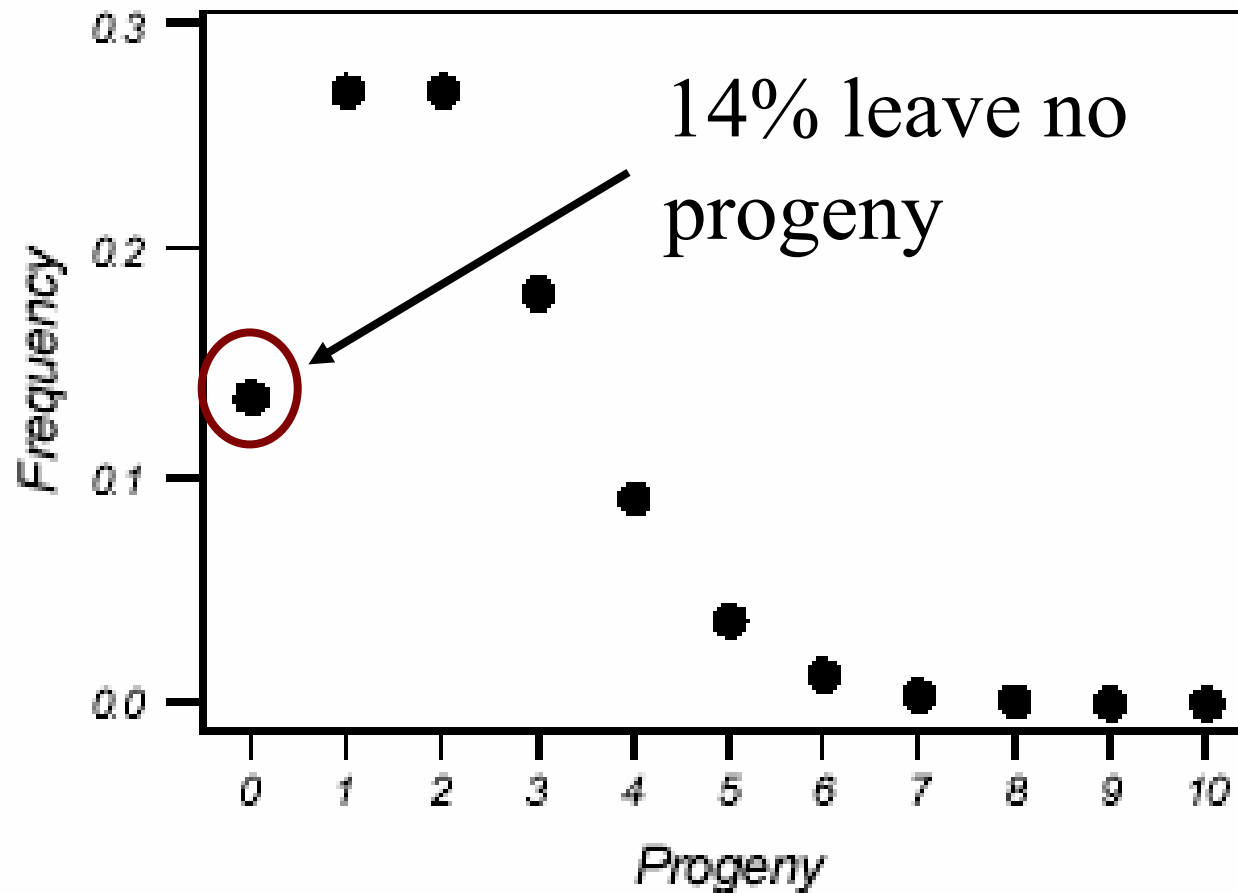
$$N_e = \frac{4 N_f N_m}{N_f + N_m}$$

Non-Random Number of Progeny

(variance in reproductive success, V_k)

$$N_e = \frac{4N - 2}{2 + V_k}$$

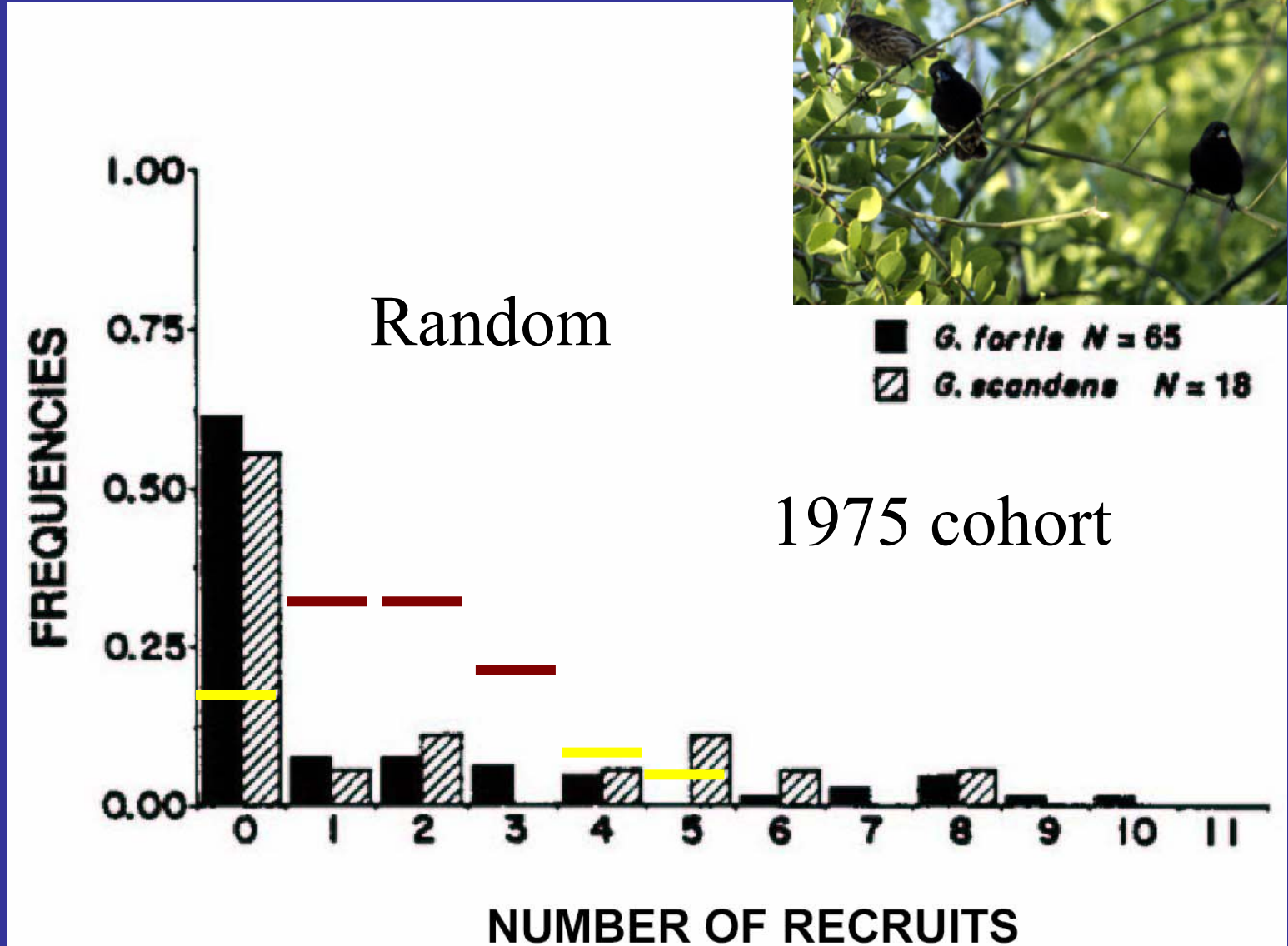
Question: What proportion of individuals do not leave any progeny in an ideal population?



Random reproductive success

Galapagos finches

Ecology 73:766-784 (1992)



Fluctuating Population Size

$$\frac{1}{N_e} = \frac{1}{t} \left(\frac{1}{N_1} + \frac{1}{N_2} + \frac{1}{N_3} + \dots + \frac{1}{N_t} \right)$$

$$N_e = \frac{t}{\sum \left(\frac{1}{N_i} \right)}$$

Reduction in N_e

(1) Unequal numbers of males and females.

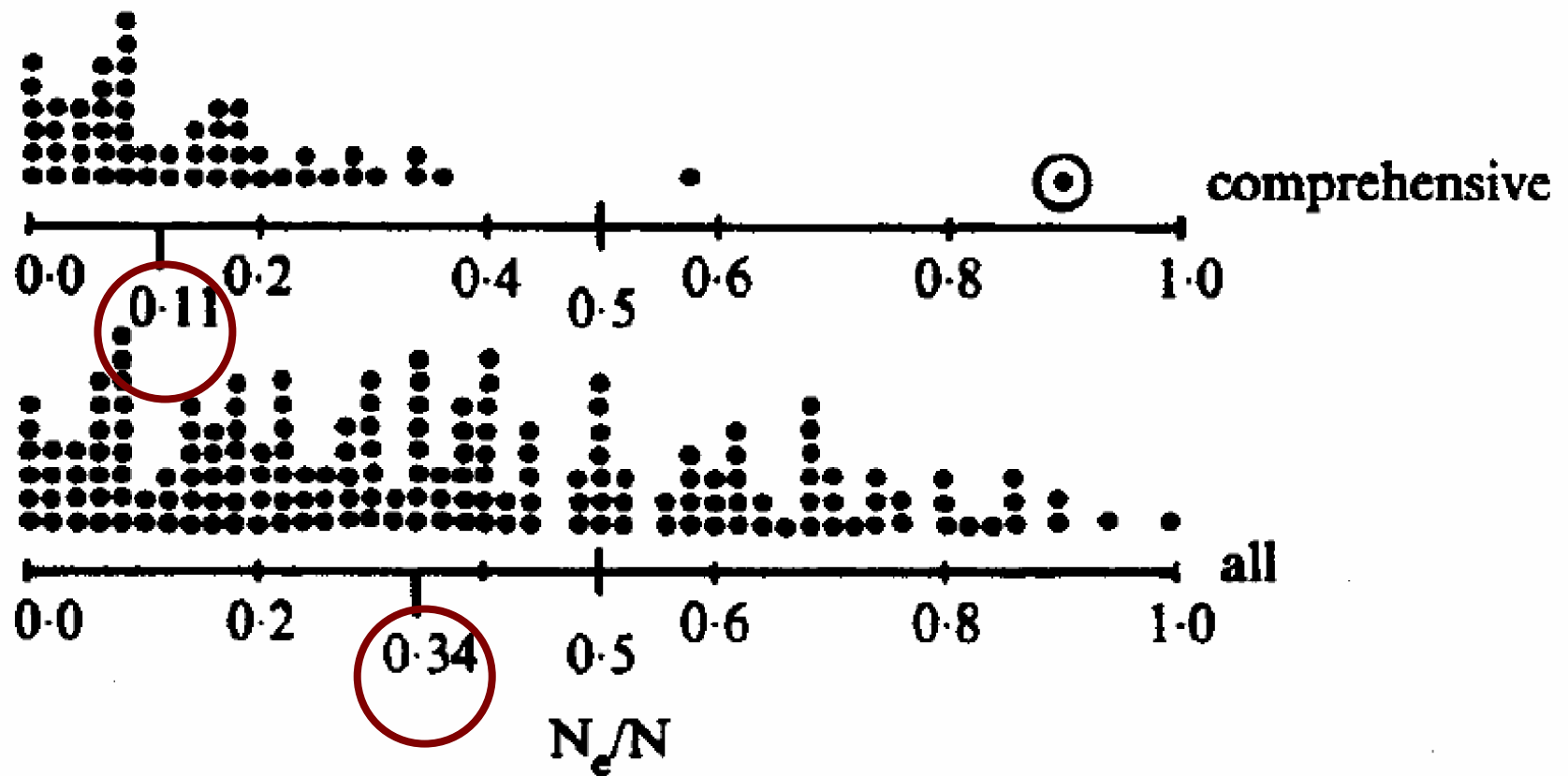
(2) Non-random reproductive success.***

(3) Fluctuating population size.***

(4) Non-overlapping (discrete) generations.

*** most important

What is N_e/N in natural populations?



Frankham, R. 1995. Effective population size/adult population size ratios in wildlife a review. *Genet. Res. Camb.* 66:95-107.

A PREVIEW: How large is large enough?

As a general rule, an N_e of 50 is necessary in the short-term to prevent immediate harmful effects of inbreeding, and an N_e of about 500 is necessary to retain enough genetic variation in order to maintain long-term evolutionary potential.

50/500 Rule

(Somewhat controversial)